Opis choroby *

Definicja

Orofaciodigital syndrome type 11 is an extremely rare, sporadic form of Orofaciodigital syndrome (OFDS; see this term) with only a few reported cases, and characterized by facial (blepharophimosis, bulbous nasal tip, broad nasal bridge, downslanting palpebral fissures and low set ears) and skeletal (post-axial polydactyly and fusion of vertebrae) malformations along with severe intellectual disability, deafness and congenital heart defects.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
OFD11
OFD11
Zespół ustno-twarzowo-palcowy, typ Gabrielli
Oral-facial-digital syndrome type 11
Oral-facial-digital syndrome, Gabrielli type
Orofaciodigital syndrome, Gabrielli type

Kod ORPHA
141000

Kod OMIM
612913

Kod ICD10
Q87.0

Kod ICD11
LD25.00

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*Źródło

orphanet